



HNF1B gene

HNF1 homeobox B

Normal Function

The *HNF1B* gene provides instructions for making a protein that attaches (binds) to specific regions of DNA and regulates the activity of other genes. On the basis of this role, the protein produced from the *HNF1B* gene is called a transcription factor. The HNF1B protein is part of a large group of transcription factors called homeodomain proteins. The homeodomain is a region of the protein that allows it to bind to DNA.

The HNF1B protein is found in many organs and tissues, including the lungs, liver, intestines, pancreas, kidneys, reproductive system, and urinary tract. Researchers suspect that this protein may play a role in the development of many of these parts of the body.

Health Conditions Related to Genetic Changes

17q12 deletion syndrome

17q12 deletion syndrome is a condition that results from the deletion of a small piece of chromosome 17 in each cell. Signs and symptoms of 17q12 deletion syndrome can include abnormalities of the kidneys (particularly fluid-filled sacs, called cysts, in the kidneys) and a form of diabetes called maturity-onset diabetes of the young type 5 (MODY5). The combination of kidney cysts and MODY5 is sometimes referred to as renal cysts and diabetes (RCAD) syndrome. Other features of 17q12 deletion syndrome include abnormalities of the urinary tract and reproductive system, delayed development, intellectual disability, and behavioral or psychiatric disorders. The health problems associated with 17q12 deletion syndrome vary widely, even among affected members of the same family.

The part of chromosome 17 that is deleted is on the long (q) arm of the chromosome at a position designated q12. This region of the chromosome contains at least 15 genes, including *HNF1B*. A deletion of this region results in a loss of one copy of the *HNF1B* gene in each cell, leading to a reduced amount of HNF1B protein. A shortage of this protein likely disrupts the regulation of genes that are necessary for the normal development of several organs, including the kidneys and pancreas. Studies suggest that a loss of one copy of the *HNF1B* gene underlies the kidney and urinary tract abnormalities, as well as MODY5, in people with 17q12 deletion syndrome.

prostate cancer

other disorders

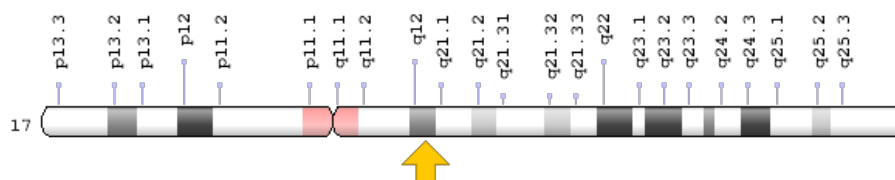
Mutations within the *HNF1B* gene can also cause abnormalities of multiple organ systems. Some of the features associated with *HNF1B* gene mutations are the same as those of 17q12 deletion syndrome (described above), including RCAD syndrome and abnormalities of the urinary tract, reproductive system, and other organs. Like the signs and symptoms of that syndrome, the health problems associated with *HNF1B* gene mutations vary widely among affected individuals. However, unlike 17q12 deletions, mutations in the *HNF1B* gene have not been found to cause delayed development, intellectual disability, or behavioral or psychiatric disorders.

Almost 200 mutations in the *HNF1B* gene have been identified. Many of these mutations change single protein building blocks (amino acids) in the HNF1B protein. Others lead to an abnormally shaped protein or prevent the production of any functional protein from one copy of the gene. A shortage of functional HNF1B protein likely disrupts the regulation of genes that are necessary for the normal development of several organs. It is unclear why these mutations can affect different organ systems in different people.

Chromosomal Location

Cytogenetic Location: 17q12, which is the long (q) arm of chromosome 17 at position 12

Molecular Location: base pairs 37,686,431 to 37,745,078 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- FJHN
- hepatocyte nuclear factor 1B
- HNF-1-beta
- HNF-1B
- HNF1 beta A

- HNF1beta
- HNF2
- homeoprotein LFB3
- HPC11
- LF-B3
- LFB3
- TCF-2
- TCF2
- transcription factor 2, hepatic
- VHNF1

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Homeodomain Proteins Constitute a Special Class of Helix-Turn-Helix Proteins
<https://www.ncbi.nlm.nih.gov/books/NBK26806/#A1240>
- RareRenal: HNF1B – Patient Information
<http://rarerenal.org/patient-information/hnf1b-patient-information/>
- The Genetic Landscape of Diabetes (2004): MODY5: Caused by a Mutation in Transcription Factor TCF2 (HNF1B)
<https://www.ncbi.nlm.nih.gov/books/NBK1666/#A891>

GeneReviews

- 17q12 Recurrent Deletion Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK401562>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28HNF1B%5BTIAB%5D%29+OR+%28HNF1+homeobox+B%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

OMIM

- HNF1 HOMEBOX B
<http://omim.org/entry/189907>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_HNF1B.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=HNF1B%5Bgene%5D>
- HGNC Gene Family: HNF class homeoboxes
<http://www.genenames.org/cgi-bin/genefamilies/set/524>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11630
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6928>
- UniProt
<http://www.uniprot.org/uniprot/P35680>

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